

Children at risk after sperm donor develops late onset genetic disease

Tony Sheldon *Utrecht*

A Dutch hospital has informed parents of 18 children conceived through artificial insemination that the children have a 50% chance of developing autosomal dominant cerebellar ataxia, inherited from affected donor sperm.

The hospital had wrestled for three years with the information, knowing that the particular subtype of the disorder can be neither identified in a carrier before symptoms appear nor properly treated.

The issue has sparked a debate over the right of children and parents to be informed in such cases and the screening of donors' gametes.

The man carrying the hereditary disorder had donated sperm

from 1989 to 1995 to the Jeroen Bosch Hospital in Den Bosch for its artificial insemination programme. The current guidelines on screening for HIV, hepatitis C, syphilis, and chlamydia and any known hereditary disorders in the family had been followed carefully.

Neither the donor nor the hospital had any indication that he carried a hereditary disorder. Symptoms of the progressive brain disorder, beginning with speech and mobility problems, usually appear between the ages of 20 and 50.

Two years later, in 1997, the donor began to suspect a problem and informed the hospital, which destroyed his remaining frozen sperm and informed the

healthcare inspectorate. Once his condition was confirmed the hospital began a lengthy process of consulting medical and ethical experts on whether and how to inform parents. In October 2001 the hospital decided to tell the parents through their general practitioners and to offer counselling and support.

The hospital has been criticised for taking too long to take a decision, but it argues that it needed the time to consult and to consider carefully the questions raised. The chairman of the hospital's management board, Frans Croonen, said the hospital took three years to resolve the "terrible dilemma" because it would be "denying people so much joy and giving uncertainty in return."

The decision was taken also because the children, aged 7 to 13, would shortly reach the age of reproduction and would then be able to pass on the disorder. However, in the future technolo-

gy may be able to confirm whether people carry the particular subtype of this disorder.

An ethicist at Maastricht University, Guido de Wert, said the hospital faced a genuine dilemma: "It was a basket of bad apples. The advantage of openness is that the child can later make an informed decision on reproduction. The disadvantage is that the news is very threatening, as there are no preventive or therapeutic options."

He added that there is also the consideration that parents might not yet have told the children about their genetic origins. Though three years was a long time, he argues that the hospital made a "defensible decision."

Pim Janssens, chairman of the association of sperm banks, rejected broader DNA screening as impractical, too costly, and raising further ethical questions about the extent of screening and requirements to inform the donor. □

Patients with emergency brain disorders are denied specialist care

Annabel Ferriman *BMJ*

The number of consultant neurologists needs to expand from the present 350 to between 1000 and 1400 if Britain is to have a comprehensive service for all acute neurological emergencies in adults, a new report says.

Patients who are critically ill should be seen immediately, and all other acutely ill patients should be seen by a neurologist within 24-48 hours, the report from the Association of British Neurologists says.

At present many patients who are acutely ill with neurological conditions such as epilepsy, Guillain-Barré syndrome, and stroke may not see a neurologist for several days, if at all.

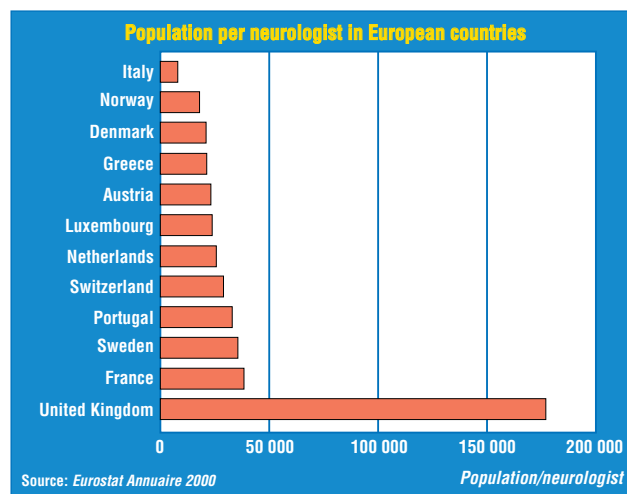
Yet when such patients are seen by a neurologist the diagnosis is often changed or a new one made. A study published in 1996 showed that of 169 patients seen by a neurologist as ward referrals 18% had their diagnosis changed. Seven patients had their diagnosis of epilepsy reversed, one had his diagnosis changed from dementia to phenytoin toxicity, and 15% of the patients were given new diag-

noses, not having been given one previously (*Journal of Neurology, Neurosurgery, and Psychiatry* 1996;61:653-4).

The association believes that some patients, such as those with status epilepticus, sudden coma, possible meningitis, or possible subarachnoid haemorrhage, need to be assessed immediately by a neurologist.

But this is only a realistic proposition in hospitals that have a neurology centre or a neurology and neurosurgery centre. Britain has only about 35 such hospitals, and more than 200 district general hospitals do not have a 24 hour, on-call, on-site neurology service. In most of these hospitals the neurology service is delivered by visiting neurologists from elsewhere.

The association thinks that a proper 24 hour emergency service can be provided only if every district general hospital has five consultant neurologists, or if every such hospital is provided with the services of three consultant neurologists who are based during the working week at a local neurology centre that also



has an emergency system outside working hours.

Each of Britain's 35 neurology and neurology and neurosurgery centres would also need at least five neurologists to carry out its work.

Using these numbers, the association has calculated two possible totals for the number of consultants required. It thinks Britain will require about 1400 consultants if emergency services are to be based at district general hospitals or about 1000 if services are to be run from neurology centres.

It says that there is not a single correct method for reaching these numbers. To reach the

higher total in 10 years the rate of growth in the number of consultants would have to rise from its current level of 7% a year to 18% a year, it says.

But even if the number of neurologists were to rise to 1400, the ratio of consultants to the UK population would still only be 1:43 000, considerably less than levels in the rest of Europe of 1:8000 to 1:38 000. □

Acute Neurological Emergencies in Adults 2002 is available free of charge from the Association of British Neurologists (tel 020 7405 4060, fax 020 7405 4070, abn@theabn.org)